



## ANTXR2 gene

anthrax toxin receptor 2

### Normal Function

The *ANTXR2* gene provides instructions for making a protein called anthrax toxin receptor 2 (ANTXR2). This protein is involved in the formation of tiny blood vessels (capillaries). Researchers believe that the ANTXR2 protein is also important for maintaining the structure of basement membranes, which are thin, sheet-like structures that separate and support cells in many tissues. This protein is called anthrax toxin receptor 2 because it allows the toxin that causes anthrax to attach to cells and trigger disease.

### Health Conditions Related to Genetic Changes

#### infantile systemic hyalinosis

At least 10 mutations in the *ANTXR2* gene have been shown to cause infantile systemic hyalinosis. These mutations most likely disrupt the formation of basement membranes, allowing a clear (hyaline) substance to leak through and accumulate in various parts of the body.

#### juvenile hyaline fibromatosis

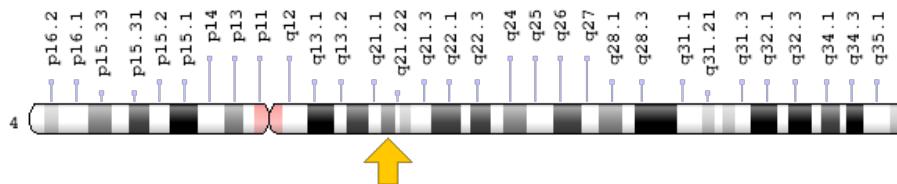
Approximately 10 mutations in the *ANTXR2* gene have been shown to cause juvenile hyaline fibromatosis. These mutations also lead to the production of an abnormally functioning ANTXR2 protein that is believed to disrupt basement membrane formation.

It is not known why some mutations in this gene cause infantile systemic hyalinosis and others cause the less severe juvenile hyaline fibromatosis. Some research has shown that the mutations responsible for these two diseases tend to occur in different regions of the gene.

## Chromosomal Location

Cytogenetic Location: 4q21.21, which is the long (q) arm of chromosome 4 at position 21.21

Molecular Location: base pairs 79,901,617 to 80,073,472 on chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- ANTR2\_HUMAN
- capillary morphogenesis protein 2
- CMG-2
- CMG2
- FLJ31074
- ISH
- JHF
- MGC111533
- MGC45856

## Additional Information & Resources

### GeneReviews

- Hyalinosis, Inherited Systemic  
<https://www.ncbi.nlm.nih.gov/books/NBK1525>

## Scientific Articles on PubMed

- PubMed

<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ANTXR2%5BTIAB%5D%29+OR+%28anthrax+toxin+receptor+2%5BTIAB%5D%29%29+OR+%28%28CMG2%5BTIAB%5D%29+OR+%28CMG-2%5BTIAB%5D%29+OR+%28capillary+morphogenesis+protein+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

## OMIM

- ANTHRAX TOXIN RECEPTOR 2  
<http://omim.org/entry/608041>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_ANTXR2.html](http://atlasgeneticsoncology.org/Genes/GC_ANTXR2.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ANTXR2%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=21732](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=21732)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/118429>
- UniProt  
<http://www.uniprot.org/uniprot/P58335>

## **Sources for This Summary**

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Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/ANTXR2>

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